

MEASURING CFTR CYSTIC FIBROSIS SECOND STAGE

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MEDICAL BIOCHEMISTRY

CFTR Protein: Not Just a Chloride Channel?

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What is it? a ductal protein that transports Cl and HCO₃ across epithelial cells. it regulates salt balance and mucus fluidity. it is found in the lungs, pancreas

intestines, and sweat glands. Found in whom? it is found in healthy individuals, and found in children and adults. it is present from birth. in cystic fibrosis patients, it is

defective or poorly functioning. it is not measured in the blood, but present on the cell surface.



Genetic Expression and Structure:

it is produced through the expression of a gene called CFTR, which is located on chromosome 7.

the process is as follows: Gene \rightarrow mRNA \rightarrow translated into \rightarrow integrated into the cell membrane.

(the protein structure includes domains such as MSD1, MSD2, ECLs, ICLs, NBD1, NBD2, and RD).



Methods of Measuring CFTR :

the essential (most important) test is the Sweat Chloride Test, using the Pilocarpine iontophoresis technique. we measure the chloride concentration in sweat.

Values (same values for children and adults): * Normal: <30 mmol/L.

Borderline: 30-59 mmol/L.

Pathological (CF): ≥ 60 mmol/L.

(researchers mentioned: Paul di Sant'Agnes, Gibson and Cooke).



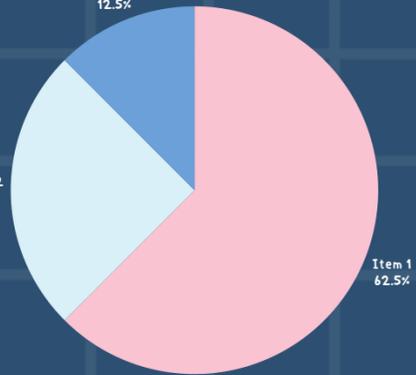
Advanced Tests and Physiological Impact:

advanced tests include Nasal Potential Difference (NPD) + measurement of ion motility in the nose

and Intestinal Current Measurement (ICM) which is a measurement from intestinal biopsy. another method is the Genetic Test for detection of CFTR Gene Mutations.

What happens if CFTR doesn't work?

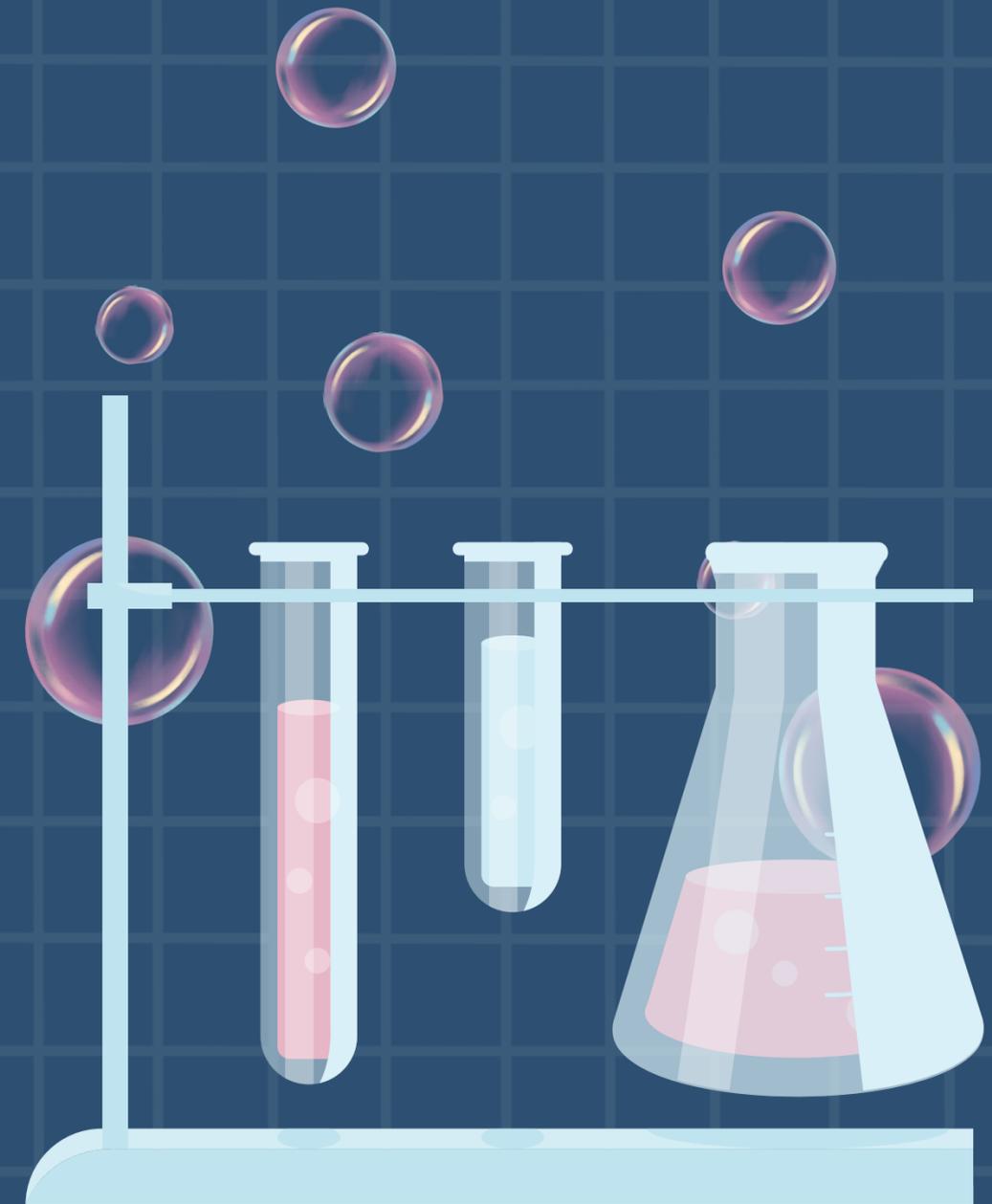
it leads to issues with chloride excretion and sodium uptake, resulting in thick mucus, chronic lung infections, and pancreatic problems.



Estimated Normal Values of CFTR Protein Concentration by Age:

**Infants (0-2 years): expected range:
not typically quantified; normal
expression should be present in
significant amounts in epithelial
tissues.**

**Children (3-12 years): expected range:
approximately 70-100 pg / μ g of
protein (varies by tissue type).**





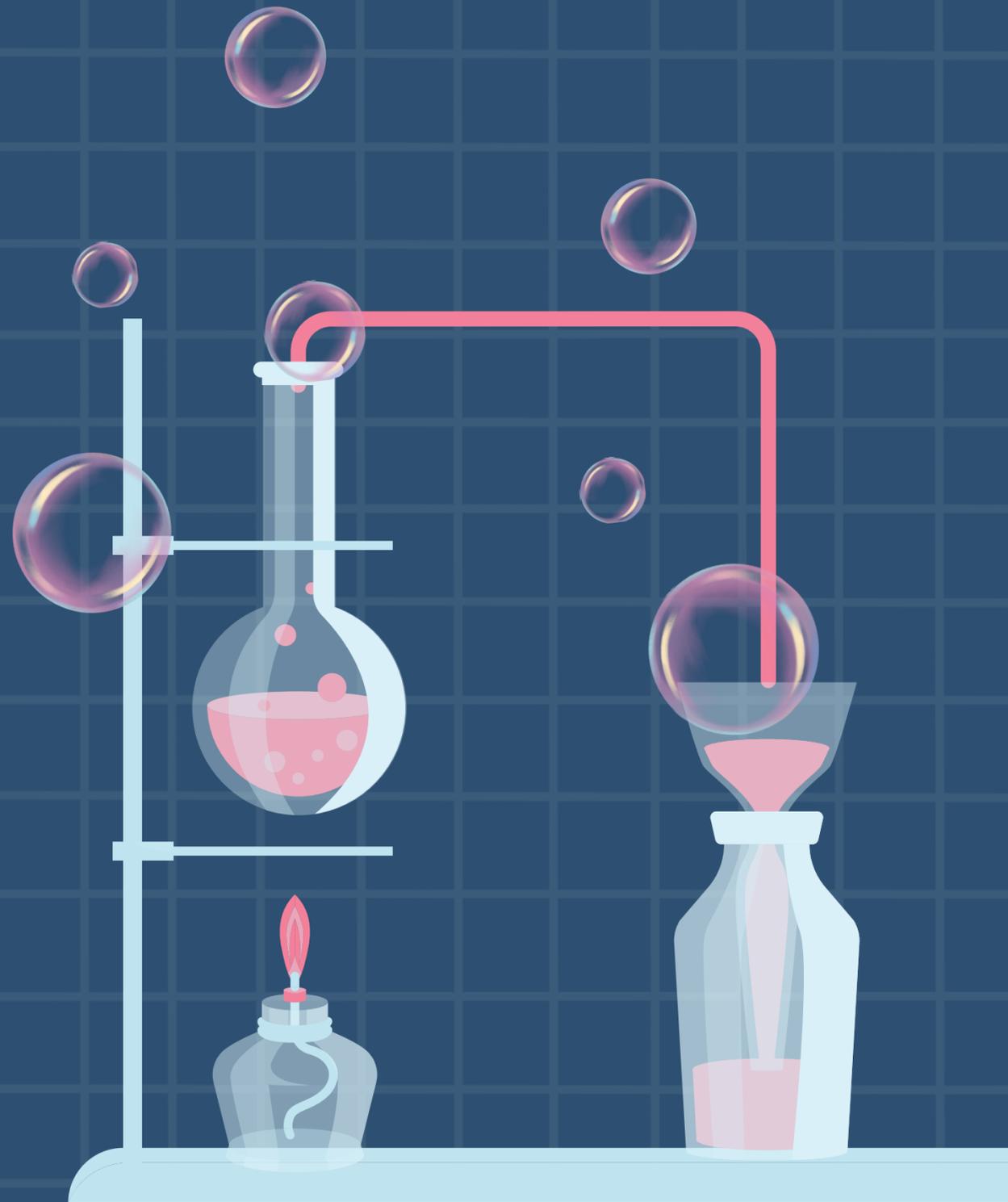
Adolescents (13-18 years): expected range: similar to adults, around 80-120 pg / μ g of protein in epithelial tissues such as the airway or gastrointestinal tract.

Adults (19+ years): expected range: about 100-150 pg / μ g of protein, depending on the tissue and individual health status.

Variability: concentrations may vary depending on the individual, specific tissue type, and method of measurement.

Measurement Methods: techniques like Western blotting or immunohistochemistry are commonly used to quantify CFTR protein levels.





**Thank
You for your
attention!**